CURRICULUM VITAE

FRANCESCO PEZZINI

Personal Information

Date of birth: 06/07/1981

Birthplace: Villafranca di Verona (VR), Italy

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Education

2010: **PhD Program in Neuroscience**; Dept. of Neurological and Movement Sciences, University of Verona, Italy;

2005: **Master's degree in Medical Biotechnologies** (Magna cum laude); Dept. of Hygiene, Microbiology and Biostatistic Sciences, University of Modena and Reggio Emilia, Italy;

2003: **Degree in Biotechnologies** - medical curriculum (Magna cum laude); Dept. of Hygiene, Microbiology and Biostatistic Sciences, University of Modena and Reggio Emilia, Italy

Positions

2010 – today: Post-Doctoral Fellow; Dept. of Neuroscience, Biomedicine and Movement Sciences (former Dept. of Neurological and Movement Sciences), University of Verona, Italy;

2007 – 2009: PhD student; Dept. of Neurological and Movement Sciences, University of Verona, Italy;

2006: Junior Research Fellow; Dept. of Neurological and Movement Sciences, University of Verona, Italy

Research Activity

Dr. Pezzini has been mainly involved in the study of genetically determined, early onset degenerative disorders affecting the Central Nervous System, which are characterized by progressive cytoplasmic accumulation of lysosomal material. The main goal of the research activity has been to study the pathogenic mechanisms involved in Neuronal Ceroid Lipofuscinosis (NCL), a group of neurodegenerative diseases marked by lysosomal accumulation of autofluorescent material with diverse composition, and by a severe neuronal cell death.

The research activity has been achieved using different methodologies:

- preparation of cell cultures, both human fibroblasts derived from skin biopsies of patients affected by different forms of NCL, and human neuroblastoma cell lines derived from SH-SY5Y cells which were engineered to alter the expression of NCL genes (either by over-expression, or by genome editing with CRISPR/Cas9 technology);
- setting up of differentiation media to get neuronal-like cells from undifferentiated neuroblastoma cells;

- gene expression analysis by RNAseq of these generated cell models;
- bioinformatics mining of gene (and protein) expression datasets to create molecular networks and to predict the activation state of specific biological functions and signalling pathways;
- cell biology techniques (immunofluorescence, fluorescent intravital traces, electron microscopy) as well as biochemical and molecular techniques (Real-Time PCR, Western blotting, enzymatic activity assay) to validate the most relevant cellular functions and the molecular networks identified through the bioinformatic inquiry.

With the combined use of these methodologies he investigated:

i) the morphology and the functionality of the endo-lysosomal compartment and of the mitochondrial reticulum;

ii) the synaptic compartment, and the expression of palmitoylated proteins, in cellular models of CLN1;

iii) the autophagic process on patients' fibroblasts affected by some genetically defined NCL forms, as well as the susceptibility of the same cell lines to various cell death mechanisms (i.e. the caspase-mediated apoptotic pathway), both under basal conditions and following specific stimuli.

The research activity is documented by scientific publications on international peer-reviewed journals and by contributions presented at both national and international meetings.

Moreover, in parallel to the research activity, Dr. Pezzini is involved in the isolation of fibroblast cultures from skin biopsies of human patients, which are then stored in a cell repository of the Neuropathology laboratory (mainly in collaboration with Dr. Bordugo of UOS Hereditary Metabolic Diseases of the AOUI Verona).

Collaborations

• Prof. Maciej Lalowski, Medicum, Biochemistry/Developmental Biology, Meilahti Clinical Proteomics Core Facility, University of Helsinki, Helsinki, Finland

• Dr. Filippo Maria Santorelli, Molecular Medicine Unit, IRCCS Stella Maris, Calambrone-Pisa, Italy

• Prof. Gian Carlo Demontis, Department of Pharmacy, University of Pisa, Italy

• Prof. Massimo Delledonne, Department of Biotechnology, University of Verona, Italy

• Dr. Bordugo, Hereditary Metabolic Diseases Unit, Paediatric Department, AOUI Verona, Italy;

Publications

• Pezzini F, Bianchi M, Benfatto S, Griggio F, Doccini S, Carrozzo R, Dapkunas A, Delledonne M, Santorelli FM, Lalowski MM, and Simonati A (2017). The Networks of Genes Encoding Palmitoylated Proteins in Axonal and Synaptic Compartments Are Affected in PPT1 Overexpressing Neuronal-Like Cells. Front. Mol. Neurosci. 10: 266. DOI: 10.3389/fnmol.2017.00266.

• Magrinelli F, Pezzini F, Moro F, Santorelli FM, and Simonati A (2017). Diagnostic methods and emerging treatments for adult neuronal ceroid lipofuscinoses (Kufs disease). Expert Opin. Orphan Drugs 5: 487–501.

• Simonati A, Williams RE, Nardocci N, Laine M, Battini R, Schulz A, Garavaglia B, Moro F, Pezzini F, and Santorelli FM (2017). Phenotype and natural history of variant late infantile ceroid-lipofuscinosis 5. Dev. Med. Child Neurol. 59: 815–821.

• Pezzini F, Bettinetti L, Di Leva F, Bianchi M, Zoratti E, Carrozzo R, Santorelli FM, Delledonne M, Lalowski M, and Simonati A (2017). Transcriptomic Profiling Discloses Molecular and Cellular Events Related to Neuronal Differentiation in SH-SY5Y Neuroblastoma Cells. Cell. Mol. Neurobiol. 37: 665–682.

• Tikka S, Monogioudi E, Gotsopoulos A, Soliymani R, Pezzini F, Scifo E, Uusi-Rauva K, Tyynelä J, Baumann M, Jalanko A, Simonati A, and Lalowski M (2016). Proteomic Profiling in the Brain of CLN1 Disease Model Reveals Affected Functional Modules. Neuromolecular Med. 18: 109–33.

• Doccini S, Sartori S, Maeser S, Pezzini F, Rossato S, Moro F, Toldo I, Przybylski M, Santorelli FM, and Simonati A (2016). Early infantile neuronal ceroid lipofuscinosis (CLN10 disease) associated with a novel mutation in CTSD. J. Neurol. 263: 1029–1032.

• Scifo E, Szwajda A, Soliymani R, Pezzini F, Bianchi M, Dapkunas A, Dębski J, Uusi-Rauva K, Dadlez M, Gingras A-C, Tyynelä J, Simonati A, Jalanko A, Baumann MH, and Lalowski M (2015). Proteomic analysis of the palmitoyl protein thioesterase 1 interactome in SH-SY5Y human neuroblastoma cells. J. Proteomics 123: 42–53.

• Scifo E, Szwajda A, Soliymani R, Pezzini F, Bianchi M, Dapkunas A, Dębski J, Uusi-Rauva K, Dadlez M, Gingras A-C, Tyynelä J, Simonati A, Jalanko A, Baumann MH, and Lalowski M (2015). Quantitative analysis of PPT1 interactome in human neuroblastoma cells. Data Br. 4: 207–216.

• Di Fabio R, Moro F, Pestillo L, Meschini MC, Pezzini F, Doccini S, Casali C, Pierelli F, Simonati A, and Santorelli FM (2014). Pseudo-dominant inheritance of a novel CTSF mutation associated with type B Kufs disease. Neurology 83: 1769–1770.

• Moro F, Gismondi F, Pezzini F, Santorelli FM, and Simonati A (2014). Clinical, ultrastructural, and molecular studies in a patient with Kufs disease. Neurol. Sci. 35: 605–607.

• Simonati A, Pezzini F, Moro F, and Santorelli FM (2014). Neuronal Ceroid Lipofuscinosis: The Increasing Spectrum of an Old Disease. Curr. Mol. Med. 14: 1043–1051.

• Santorelli FM, Garavaglia B, Cardona F, Nardocci N, Bernardina BD, Sartori S, Suppiej A, Bertini E, Claps D, Battini R, Biancheri R, Filocamo M, Pezzini F, and Simonati A (2013). Molecular epidemiology of childhood neuronal ceroid-lipofuscinosis in Italy. Orphanet J. Rare Dis. 8: 19.

• Pezzini F, Gismondi F, Tessa A, Tonin P, Carrozzo R, Mole SE, Santorelli FM, and Simonati A (2011). Involvement of the mitochondrial compartment in human NCL fibroblasts. Biochem. Biophys. Res. Commun. 416: 159–164.

• Cannelli N, Garavaglia B, Simonati A, Aiello C, Barzaghi C, Pezzini F, Cilio MR, Biancheri R, Morbin M, Bernardina BD, Granata T, Tessa A, Invernizzi F, Pessagno A, Boldrini R, Zibordi F, Grazian L, Claps D, Carrozzo R, et al. (2009). Variant late infantile ceroid lipofuscinoses associated with novel mutations in CLN6. Biochem. Biophys. Res. Commun. 379: 892–897.

• Simonati A, Cannelli N, Pezzini F, Aiello C, Bianchi M, Tessa A, and Santorelli FM (2009). Neuronal ceroid lipofuscinoses: many players, and more to come. Future Neurol. 4: 601–616.

• Cermelli C, Cenacchi V, Beretti F, Pezzini F, Luca D Di, and Blasi E (2006). Human herpesvirus-6 dysregulates monocyte-mediated anticryptococcal defences. J. Med. Microbiol. 55: 695–702.

• Moschettini D, Franceschini R, Vaccaro NM, Cermelli C, Pezzini F, Balestrieri M, Cerase A, Bartalini S, Ulivelli M, Tosi GM, and Donati D (2006). Human herpesvirus-6B active infection associated with relapsing bilateral anterior optic neuritis. J. Clin. Virol. 37: 244–247.

• Blasi E, Mucci A, Neglia R, Pezzini F, Colombari B, Radzioch D, Cossarizza A, Lugli E, Volpini G, Del Giudice G, and Peppoloni S (2005). Biological importance of the two Toll-like receptors, TLR2 and TLR4, in macrophage response to infection with Candida albicans. FEMS Immunol. Med. Microbiol. 44: 69–79.

Meeting Attendance and Contributions

• 16th International Conference on Neuronal Ceroid Lipofuscinoses (Batten Disease), London, UK, September 12nd-16th 2018. “Over-expression of CLN1/PPT1 dysregulates networks of genes coding palmitoylated proteins of axonal and synaptic compartments in neuronal-like cells”.

• Joint Meeting Congress AINPeNC – AIRIC, Milano, Italy, May 17th-19th 2018. “Networks of genes encoding palmitoylated proteins in axonal and synaptic compartments are affected in PPT1 overexpressing neuronal-like cells”. Oral Communication.

• INN Open Neuroscience Forum, Verona, July 21th 2017. “Networks of palmitoylated proteins of axonal and synaptic compartments in CLN1/PPT1 overexpressing neuroblastoma cells”. Oral Communication.

• Joint Meeting 51th Congress AINPeNC – 41th Congress AIRIC, Verona, Italy, June 4th-6th 2015. “Elongation of neuronal processes is affected in an in vitro cellular model of CLN1 disease”. Oral Communication.

• DEM-CHILD General Assembly Meeting in Hamburg, Germany, 1st-3rd September 2014. “WP5 – final report of UNIVR/Italy Unit”. Oral Communication.

• Qiagen-Ingenuity User Group Meeting & IPA New User Training, Organized by Qiagen Redwood City, Paris, France, June 19th-20th 2014.

• Joint Meeting 50th Congress AINPeNC – 40th Congress AIRIC, Verbania, Italy, June 5th-7th 2014. “Evidences of Autophagy in CLN1 Disease”. Oral Communication.

• DEM-CHILD General Assembly Meeting in Helsinki, Finland, September 2nd-4th 2013. “WP5 Task 2: to use patient cell lines and cultured models to study the cell pathology caused by Mediterranean CLN1 mutations”. Oral Communication.

• Joint Meeting 49th Congress AINPeNC – 39th Congress AIRIC – 1st Corso Residenziale in Neurogenetica, Pisa, Italy, June 29th-1st 2013. “Differentiated neuroblastoma cells featuring mature neurons: an in vitro system suitable for the investigation of neurodegenerative disorders of childhood”.

• DEM-CHILD General Assembly Meeting in Verona, Italy, October 18th-20th 2013

• Joint Meeting 48th Congress AINPeNC – 38th Congress AIRIC, Napoli, Italy, May 24th-26th 2012. “Differentiation of neuroblastoma cells: putative in vitro models to investigate childhood neurodegeneration”. Oral Communication.

• 13th International Conference on Neuronal Ceroid Lipofusinoses (Batten Disease) & Patient Organisation Meeting London, UK, March 28th-31st 2012. “Evidence of Autophagy in Human CLN6 Fibroblasts”.

• Joint Meeting XLVII Congress AINP- XXXVII Congress AIRIC, Genova, Italy May 19th-21st 2011. “Autophagy is active in human CLN6 and CLN3 fibroblasts”. Oral Communication.

• Lysosomes in health and disease – Biochemical Society, London, UK, May 13rd-14th 2010. “The endo-lysosomal system and the autophagic response in human NCL fibroblasts in vitro”.

• 12th International Congress on Neuronal Ceroid Lipofuscinoses (NCL), Hamburg, Germany, June 3rd-6th 2009.

• IV Meeting on the Molecular Mechanisms of Neurodegeneration, Milan, Italy, May 8th-10th 2009. “Mitochondrial involvement in CLN1 fibroblasts”.

• European Research Conference on Paediatric Neurology, Tubingen, Germany, October 3th-4th 2008.

• Joint Meeting XLIV Congress AINP - XXXIV Congress AIRIC, Milan, Italy, June 18th-21st 2008. “Cell Pathology of Human CLN1 Fibroblasts in vitro”.

• Joint Meeting XLIII Congress AINP - XXXIII AIRIC, Verona, Italy, September 30th-October 3rd 2007. “Cytoplasmic events in neuronal ceroid lipofuscinoses”.

• III Meeting on the Molecular Mechanism of Neurodegeneration, Milan, Italy, May 19th-21st 2007. “Does mitochondrial involvement affect fibroblast functioning in neuronal ceroid lipofuscinoses?”.

• 13th SIMMOC Congress, Modena, Italy, June 13rd-14th 2005. “Biological role of two Toll-like receptors, TLR2 and TLR4, in the macrophagic response to Candida albicans”.